UKPIPS Fact Sheet No. 036

Diagnosis Quotes

UKPIPS spent time in 2018 concentrating on the need for early diagnosis of people with a Primary Immune Deficiency. During world PID week, we called for GPs to be taught about these conditions and in particular about adult onset of CVID, during their initial GP training. These quotes illustrate exactly why early diagnosis offers a good quality of life and, by comparison, the trauma that patients and the parents of paediatric patients go through before they receive a diagnosis.

The following are quotes from the UKPIPS England Closed Facebook Group. They have all been anonymised and all contributors have given permission for them to be used by UKPIPS.

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My son was diagnosed with XLA (X linked agammaglobulinaemia, a genetic condition that only affects boys) in Dec 2016. He was discovered to have 'low' levels of immunoglobulin when tested by paediatrician looking for antibodies to cow's milk protein. The blood tests in fact showed zero levels of immunoglobulin. And we were seen at Sheffield Children's Hospital within a few days of them receiving our referral. Prior to this he had been constantly poorly from age 3 weeks with, among many others, infective eczema, diarrhoea and respiratory infections including a nasty bout of pneumonia with partial lung collapse. At each GP and hospital visit and course after course of antibiotics we questioned the function of his immune system, but this was always brushed aside, and the immediate symptoms dealt with only. Since starting antibody replacement therapy 16 months ago, supplemented by a daily dose of antibiotics, he has been very stable only having a couple of episodes of respiratory infections, which have responded quickly to further antibiotics. My son is thriving with the support of an awesome team at Sheffield, but I do wonder how much more poorly he would have been and how much longer it would have taken to get diagnosed if we had not pushed for the allergy testing.

UK Primary Immune-deficiency Patient Support

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It took 9 years to get my daughter diagnosed. She was born with eczema which was well controlled till she hit teenage years. She had her first bout of cellulitis at 15 and they wanted to remove her leg from the knee as she was too young to have cellulitis, so it must be necrotizing fasciitis. Her skin was constantly dry, itchy and flaky. She was given numerous steroids and moisturisers to try. Every time she had a bout of cellulitis her skin got worse, even the dermatologists she saw asked what was wrong with her skin. Eventually a junior doctor saw her and really got his teeth into her case. After a couple of years of tests, appointments, tablets and creams and 5 days on ICU and with sepsis (because the hospital consultant didn't believe she had cellulitis) she was diagnosed with PID. She is now 2 years into immunoglobulin treatment and she is so much better. It's just a shame that diagnosis took so long. No one looked at her symptoms as a whole, it was only bits of things that were fixed. Her son is now under an immunologist and is checked yearly just in case

We had to query privately for a child ourselves as it was missed by paediatrics et al. And when I queried this with the immunologist I was pretty much laughed at. All of my child's immunoglobulins are low and they are also producing antinuclear factor antibodies. There are no specific guidelines for paediatrics and they are being seen by adult immunologists who don't have skills set to work with children. We are also being told by primary care that the tertiary service have to treat infections and vice versa by primary care. This means we have no access to NHS services. We've even been told by the CCG not to go to A&E

I am diagnosed with Good Syndrome which is a rare, adult-onset primary immunodeficiency suspected in patients who exhibit hypogammaglobulinemia and low levels of B cells along with a benign thymic tumour (thymoma). I had a thymoma removal operation and was told at the time that no further treatment was necessary. Unfortunately, that proved to be exceedingly incorrect advice. I started getting passed from GP to GP after suffering frequent opportunistic infections involving sinuses and lungs. I was signed up to an asthma clinic, subjected to numerous chest X-rays, given various antibiotics, nasal sprays etc., and most seriously hospitalised in a coma for 7 days with a case of meningitis. Generally, I witnessed a severe decline in my general health. I was eventually sent to one chest physician who had an iota of knowledge which was enough for him to send me to an immunologist. This took 10 years from my original thymoma operation. I was given a diagnosis within 5 minutes of meeting the Immunologist and I actually burst into tears with the relief that I had a very real condition and that my health could be improved with the right treatment and someone who knew what the right course of treatment should be. Sadly, I still meet GPs who have no idea about PID and my condition.

When I was a year old (20 years ago) I was diagnosed with an 'undefined immune deficiency', but this only came due to so much pushing from my parents to be tested for everything. My GP didn't believe that something was wrong with me. The doctors and nurses at the Whittington only agreed to give me a chest X-ray if my mum agreed to have psychiatric help (turned out I had an abscess on my lung!) it was only then that my parents were finally believed, and I was transferred to GOSH.

My little boy still doesn't have an immunology diagnosis - despite the fact we know he has poor MBL, IGA and IGG. He receives treatment via respiratory as his serious infections were chest related, and now has potential lung scarring. Prophylactic antibiotics

I suffer from IBS and chest infections every change of season. I was sent for bloods because of stomach pain. Came back with low IgG. I was referred to an immunologist and then put on immunoglobulin once every three weeks. It didn't agree with me and I ended up with aseptic meningitis. I was changed to subcutaneous immunoglobulin product once a week and trained for home therapy. I'm still getting chest infections and have now been diagnosed with asthma and GERD... my infusions are now on a break for 6 months to see how I cope without it and if I can manage on prophylactic antibiotics instead.

I feel there are a lot of people diagnosed with other disorders prior to CVID, which is the primary diagnosis. I was diagnosed with ITP way before my CVID diagnosis although I have been told that the ITP is an off-shoot of the CVID. I speak to a lot of people with ITP who are saying they are always ill with infections and feel that they should be tested for PID. If Doctors have patients with repeated infections they should be thinking PID quicker and looking at reasons why. Too often, PID is getting diagnosed by chance and not through good practice by health providers.

I was seeing a chest consultant privately and becoming more and more ill with pneumonia they could not get rid of. I'd asked my GP and my consultant to refer me to the London Chest Hospital, but they would not. Luckily, I did an NVQ level 2 in anatomy and physiology and discovered that my immune system was not performing as I was being taught it should. I asked my consultant to test my immune system and he more or less laughed. The next time I saw him I was very uptight and told him he had not only failed to cure me but had made me worse! He then tested my blood and found my PID. I'd been ill most of my life on and off, mainly on, but for 8 years he'd neglected to look further into matters.

My journey is mild compared to a lot of other people's. At age 63 I was referred as an emergency to Haematology with a platelet count of 6 (normal range is 150-400). I was diagnosed with ITP. After a few months of seeing the Haematology consultant he mentioned that my immunity figures were "a bit low" and he would refer me to Immunology. The next time I saw him I asked whether there had been any feedback from Immunology because I had not heard from them. He had forgotten to refer me but did so then. Thus, began my journey into diagnosis of CVID. I have been receiving IVIG for 4 years now and keep well. Having said that, I live a fairly rarefied existence - I avoid young children and everyone who has colds, flu, etc and shingles. I forgot to say that throughout my life I have suffered from appalling sinusitis, one bout of pneumonia, viral meningitis (age 19), ear infections and 3 bouts of shingles on my face.

I was diagnosed at South Shields practically straight away after rare type bacterial meningitis septicaemia.

My sister's GP failed to diagnose CVID in my sister, despite repeated chest infections and pneumonia. She ended up in hospital with pneumonia, the same hospital my Dad was being treated at for CVID. They both ended up at chest x-rays at the same time and then hospital doctors twigged that they were Father and Daughter and they might have the same thing! I also have CVID but was diagnosed completely by chance due to ITP being diagnosed first under a Haematologist, countless blood tests and no CVID diagnosis. A Doctor treating my Dad asked family members to be tested; and lo and behold, “yes, you have the same thing”. Due to late diagnosis my sister ended up with encephalitis and died and I have lung damage, deafness etc. I feel that if my sister was having repeated infections why did the doctor not dig deeper? When I had shingles at 14 surely that should have raised a red flag?

I’m certainly in a much better place thanks to a wonderful immunology team at St James’. But it does concern me that, due to ignorance there are others going through what I went through. It was only by chance that I had an appointment with the one chest specialist who referred me, I had seen plenty of others. He only referred me because he wasn’t happy with the results of a blood test he’d requested!

In retrospect I have found that immunologists didn't read their own textbooks or follow their own diagnostic tests. Before the stat 3 test, my son, James, had all the symptoms of Hyper IgE. His first diagnosis from his paediatric consultant in collusion with a Geneticist was Hyper IgE. When he was referred to an immunologist he changed his diagnosis to CMC, which led him down the route of eventually taking him off his IVIG which caused his multiple pneumonias and irreparable damage to his lungs. James had Asperger's, ADHD and XYY syndrome. Whilst on a treatment regime he responded well, had a normal childhood right up until they removed that regime (replacement immunoglobulin therapy) because of his Asperger's he wasn't compliant with the new regime. I had to watch him when giving his medication, he began to lose his appetite. He was given a PEG for over-night feeding. It remained in his stomach and he eventually threw it up. He developed scoliosis. They ignored it at first, until it got so bad they referred him to an orthopaedic surgeon, who, because of his low weight, refused corrective surgery. He was refused a lung transplant without a bone marrow transplant. He put on the weight required to have corrective surgery on his spine but never received it. The carrots were dangled and with someone living with Asperger's it led to depression… and so, the spiral began. He tried to sue, but the medical community closed up as in the traditional way. Some retired and others emigrated. His case failed, and James died. Justice was not done.

I really think the paediatric situation requires urgent attention. If that was the situation today, they would launch Fabricated illness proceedings and remove the child.

Our daughter's path to diagnosis was challenging. We were told "she would grow out of it by the time she starts school" and so from the age of 3-4 we carried on giving daily antibiotics as prevention. Watched her struggle with monthly chest infections for 2 more years, counting down the months until she would start school and she would be absolutely fine!? By the April before she started school, we could tell she was getting worse, not better. We rang the consultant's nurse and they said why did we wait so long. I still feel stupid for waiting so long, but when she was diagnosed with CVID that month, it was literally the first time we had any suggestion that she had a life-long disease. After such a challenging first few years of her life, we just hung on to the hope that she was going to grow out of it!! Thankfully, now (5 years later) my daughter has done really well with her infusion treatments and school. For us it was actually the immunology team that said she would grow out of it, and it was our wonderful respiratory consultant that made the diagnosis.

There is a lot of cross-over. Sadly, I think with immune deficiency it is all about seeing the right person at the right time.

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